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New Autosomal Recessive Syndrome of Progressive Sensorineural Hearing Loss and Cataracts: Report on Two Brazilian Patients

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We report on two sisters with cataracts and progressive sensorineural hearing loss, starting in infancy. They were born to consanguineous parents, and there were no similar cases in the family. To our knowledge this is the first report on this autosomal recessive condition. Clinical and genetic aspects are discussed. Am. J. Med. Genet. 70:247-249, 1997. © 1997 Wiley-Liss, Inc.

KEY WORDS: autosomal recessive inheritance; consanguinity; progressive sensorineural hearing loss; cataracts

INTRODUCTION

Sensorineural hearing loss is part of the clinical picture of some hundred of environmental, chromosomal, and inherited conditions [Gorlin et. al., 1995], but its association with cataracts is uncommon [Jan et al., 1976; Nadol and Burgess, 1982; Nucci and Mets, 1990; Guala et al., 1992]. Here we report on a newly recognized autosomal recessive progressive sensorineural hearing loss associated with cataracts in two sisters. Clinical and genetic aspects are discussed.

CLINICAL REPORT

Patient 1

RCS (Fig. 1A,B), the proposita, born in 1946, was the 4th child of a normal 27-year-old G4P4 mother and her first cousin, 35-year-old husband. Pregnancy was unremarkable. There was no exposure to toxic agents or X-rays and no trauma was reported. Patient was born at term, and delivery was normal. Birth measurements were not recorded. Bilateral corticonuclear asymmetric cataracts appeared in late infancy, and were removed at age 16 and 35 years. At age 14 she noted a slowly progressive hearing loss.

Clinical examination at age 50 years showed: weight 42 kg (< 3rd centile), height 1.54 m (10th centile), and head circumference 55 cm (50th centile). She presented severe sensorineural hearing loss and visual acuity only to light. There were no additional clinical signs. Results of laboratory blood tests and chromosomes were normal.

Audiological examination: Audiogram (Fig. 2) showed a 85–105 dB bilateral profound sensorioneural hearing loss. Speech discrimination was severely impaired (Speech detection: Right ear: 85 dB; Left ear: 105 dB and Speech Recognition Index: Right ear: 0%; Left ear: 0%.). Timpanometry was normal with absent reflexes. Caloric vestibular tests were normal. Evoked otoacoustic emission were absent.

Ophthalmological examination showed: esotropia, opacity of both lens, and normal intra ocular pressure (IOP).

Patient 2

MCCC (Figs. 3A,B), the younger sister of the proposita, was born in 1948. Pregnancy was unremarkable. There was no exposure to toxic agents or X-rays and no trauma was reported. Patient was born at term, and delivery was normal. Birth measurements were not recorded. Bilateral corticonuclear cataracts appeared in late infancy and were asymmetric. At age 12 she started presenting slowly progressive sensorineural hearing loss.

Clinical examination at age 48 years showed: weigth 40.7 kg (<3rd centile), heigth 1.55 m (10th centile), and head circumference 56 cm (50th centile). She presented with severe sensorineural hearing loss and visual acu-

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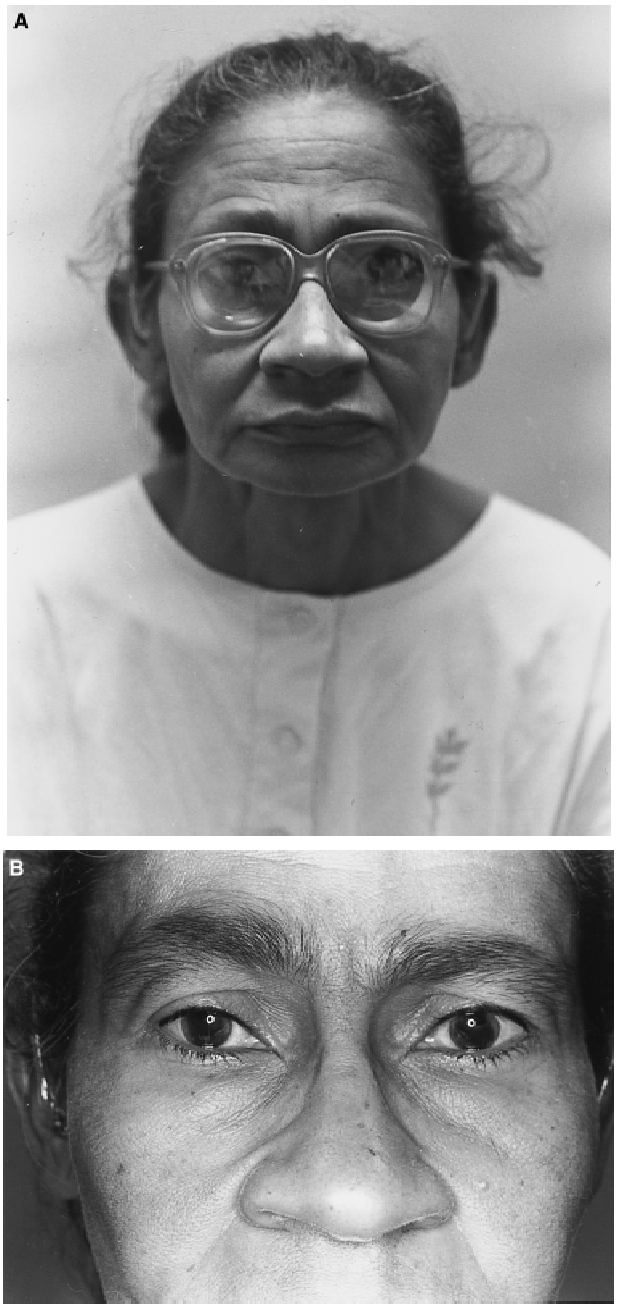

Fig. 1. **A, B:** Clinical aspects of the proposita.

ity only to light. There were no additional clinical signs. Results of laboratory blood tests and choromosomes were normal.

Audiological examination: Audiogram (Fig. 4) showed a 95–120 dB bilateral profound sensorineural hearing loss. Speech discrimination was severely impaired (Speech detection: Right ear: 100 dB; Left ear: 115 dB; Speech recognition Index: Right ear: 0%, Left ear: 0%). Timpanometry was normal with absent reflexes. Caloric vestibular tests were normal. Evoked otoacoustic emission were absent.

Ophthalmological examination showed: esotropia, opacity of both lens, and normal intra ocular pressure (IOP).

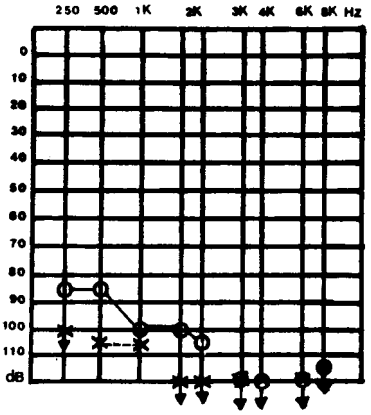

Fig. 2. Audiogram of the proposita

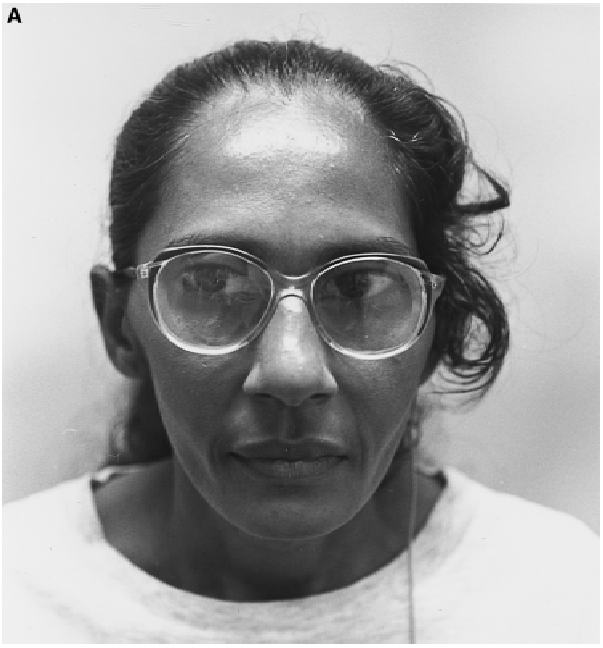

Fig. 3. **A, B:** Clinical aspects of the proposita's sister.

DISCUSSION

Sensorineural hearing loss and cataracts have been reported as part of the clinical picture of some malformative and metabolic syndromes [Guala et al., 1992; Gorlin et al., 1995]; however, the association of progressive sensorineural hearing loss and cataracts, congenital or starting in late infancy, is rare [Harboyan et al., 1971; Jan et al., 1976; Nadol and Burgess, 1982; Nucci and Mets, 1990; Guala et al., 1992].

In two instances this association was reported as an autosomal dominant trait [Nadol and Burgess, 1982; Guala et al., 1992]. The pattern of inheritance clearly distinguishes these reports from the present family. Progressive sensorineural hearing loss and cataracts have been also reported with autosomal recessive inheritance [Harboyan et al., 1971; Jan et al., 1976; Nucci and Mets, 1990]. Harboyan et al. [1971] reported on 2 sibs born to consanguineous parents (first cousin) presenting corneal dystrophy evident at birth and slowly progressive sensorineural hearing loss starting between 10 to 25 years. The time of manifestation and the type of eye involvement clearly differentiate Harboyan syndrome from the present condition. Cataracts, sensorineural hearing loss, congenital total color blindness (monochromatism) and hyperinsulinism, was reported by Jan et al. [1976]. The whole clinical picture as well the monochromatism and hyperinsulinism present in Jan's cases clearly distinguish them from the patients here reported.

Cataracts and sensorineural associated with spasticity of lower limbs, mental retardation, and hypercholesterolemia was reported by Nucci and Mets [1990]. The neurological involvement of lower limbs, the presence of mental retardation and the hypercholesterolemia are the main cardinal points of differentiation with the present syndrome.

In conclusion, the present syndrome seems to be distinct from all those reported. Normal parents, affected sisters, parental consanguinity suggest autosomal recessive inheritance.

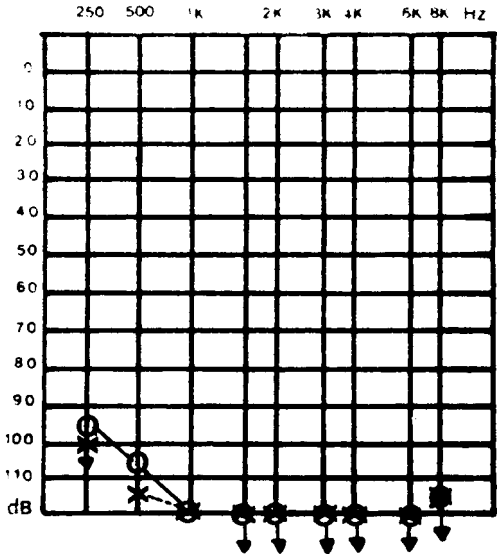

Fig. 4. Audiogram of the proposita's sister.

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